



SPGH

Sociedade Portuguesa de Genética Humana



2nd Practical course on computational tools and bioinformatics for next-generation sequencing

12th - 13th April 2019

Venue: *Departamento de Engenharia Informática, Instituto Superior de Engenharia do Porto
Rua Dr. António Bernardino de Almeida, 431, 4200-072 Porto*

Programme

Module 1: From raw data to workable files

Christian Gilissen. Department of Human Genetics, Radboud University Medical Center
(Christian.Gilissen@radboudumc.nl)

- Primary NGS bioinformatics overview
- Basics of common NGS file formats: FASTQ, BAM and VCF
- Basics of interpretation of SNV/indel
- Practical interpretation of exomes

Module 2: ENSEMBL Browser: NGS data annotation & data mining

Emily Perry. ENSEMBL, European Bioinformatics Institute (EMBL-EBI)
(emily@ebi.ac.uk)

- Introduction to ENSEMBL
- Variant Effect Prediction: Accessing variation data and variant effect prediction using Ensembl
- Data export with *BioMart* - retrieving genomic information using a web interface

Module 3: VarAFT filtering candidate variants and genes

Jean-Pierre Desvignes & David Salgado. IGE Bioinformatique, Aix-Marseille Université
(jean-pierre.desvignes@univ-amu.fr; david.salgado@univ-amu.fr)

- Introduction to VarAFT tool
- Filtering candidate variants and applying distinct inheritance disease models
- Practical examples of gene panels and exome cases



Timetable

Friday 12th

08:45h Welcome and introduction to the course

Rosário Santos & Jorge Oliveira

Friday 12th		Saturday 13th	
09:00 - 09:45h	(T) Module 1: C. Gilissen	09:00 - 10:00h	(T) Module 3: D. Salgado & J.P. Desvignes
09:45 - 10:30h	(T) Module 2: E. Perry	10:00 - 10:30h	(TP) Module 3.1: D. Salgado & J.P. Desvignes <i>Group A</i> (P) Module 2.2: E. Perry <i>Group B</i>
Coffee break		Coffee break	
11:00 - 13:00h	(TP) Module 1.1: C. Gilissen <i>Group A</i>	11:00 - 11:45h	(TP) Module 3.1: D. Salgado & J.P. Desvignes <i>Group A</i> (P) Module 2.2: E. Perry <i>Group B</i>
	(P) Module 2.1: E. Perry <i>Group B</i>	11:45 - 13:00h	(TP) Module 3.1: D. Salgado & J.P. Pierre <i>Group B</i> (P) Module 2.2: E. Perry <i>Group A</i>
13:00 - 14:00h	Lunch	13:00 - 14:00h	Lunch
14:00 - 16:00h	(TP) Module 1.1: C. Gilissen <i>Group B</i>	14:00 - 16:00h	(P) Module 3.2: J.P. Desvignes <i>Group A</i> (P) Module 3.2: D. Salgado <i>Group B</i>
	(P) Module 2.1: E. Perry <i>Group A</i>		
Coffee break		Coffee break	
16:30 - 18:00h	(P) Module 1.2: C. Gilissen <i>Group A</i> (P) Module 1.2: C. Gilissen <i>Group B</i>	16:30 - 17:30h	(P) Module 3.2: J.P. Desvignes <i>Group A</i> (P) Module 3.2: D. Salgado <i>Group B</i>

T - Lecture (auditorium)

TP - Practical demonstrations (computer rooms)

P - Practical hands-on exercises (computer rooms)